

## CASE REPORT

# Pentalogy of Cantrell: A Rare Multiple Congenital Malformation Syndrome

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## ABSTRACT

Pentalogy of Cantrell is a rare multiple congenital malformation syndrome characterized by combination of five features: abdominal wall defect, defect of lower sternum, defect of diaphragmatic pericardium, defect of anterior diaphragm and congenital cardiac anomalies. These defects can be diagnosed as early as 1st trimester of pregnancy. The complexity of these anomalies, in particular the presence of cardiac defects, determines the management as well as prognosis.

**Keywords:** Abdominal wall defect, Ectopia cordis, Pentalogy of Cantrell.

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## INTRODUCTION

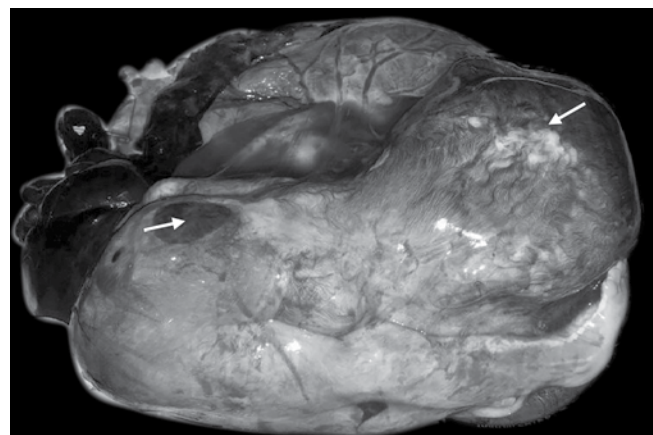
Thoracoabdominal ectopia cordis or pentalogy of Cantrell is a rare congenital syndrome of anterior abdominal wall defect, lower sternal defect, diaphragmatic defect and intracardiac abnormalities. First described by Cantrell in 1958, the syndrome occur sporadically with variable degree of expression.<sup>1,2</sup> We here present a case report of incomplete expression of pentalogy of Cantrell.

## CASE REPORT

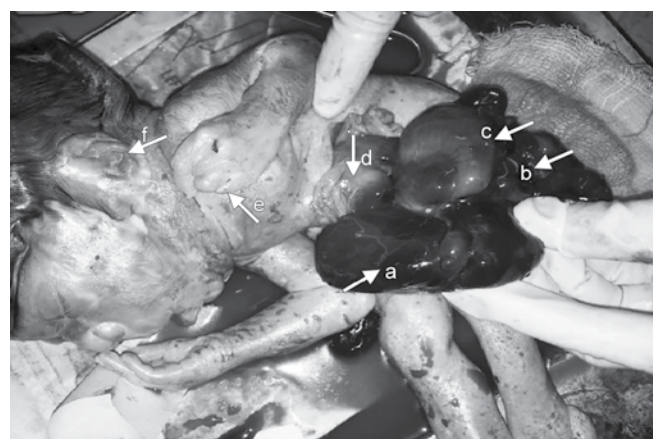
A 30-year-old unbooked female gravida 4, para 1, abortion 2, with a previous history of normal delivery, was admitted to our hospital with vide Cr number 18014 on June 4, 2013, at 31 weeks of gestation with ultrasonography (USG) showing multiple congenital anomalies. Patient was examined. Her vitals were stable. On per abdomen

examination, uterus was relaxed, 30 weeks. Patient investigated. A repeat scenography from our institute was done which showed single alive fetus with measurements corresponding to 31 weeks of gestation. The placenta was grossly normal. Polyhydramnios was present. An unusual finding was noted in thoracoabdominal region of fetus. There was defect in anterior abdominal wall and chest wall. Abdominal organs and heart were lying in amniotic cavity. Spine was grossly deformed and kyphotic. Lateral ventricles, 3rd and 4th ventricles of brain were dilated, thinning of brain parenchyma seen suggestive of hydrocephalus (Figs 1 and 2).

Patient was referred to higher center where facility of pediatric surgery was available but she refused. Patient went into spontaneous labor and a single alive male baby (birth weight 1500 gm) was delivered with small cord on June 12, 2013. Baby died immediately after birth. On



**Fig. 1:** Spinal deformity and hydrocephalus



**Fig. 2:** Pentalogy of Cantrell: (a) Liver, (b) intestine, (c) stomach, (d) heart, (e) hand deformity and (f) ear deformity

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Fig. 3: Ectopia cordis



Fig. 4: Abdominal viscera protruding out of abdominal wall defect



Fig. 5: Heart and liver lying in amniotic cavity (Hrt: Heart; Liv: Liver)



Fig. 6: Intestinal loops protruding out

examination of baby, anterior abdominal wall defect was present at the level of cord insertion. Intestinal loops, stomach, spleen, liver and heart were protruding out of defect (Figs 3 to 6). Placental weight was 200 gm with no abnormality. Autopsy was advised but relatives refused due to social issues. Postpartum period was uneventful. Our anatomical and sonographic findings correlated.

### DISCUSSION: EMBRYOGENESIS

Sternum, abdominal wall, pericardium and part of diaphragm arise from somatic mesoderm, while the myocardium arises from splanchnic mesoderm. An event occurring prior to differentiation of the mesoderm into these two layers could produce defect in all of the involved structures as seen in pentalogy of Cantrell. Although the specific etiology is unknown, the timing of the event would be between 14 and 18 days after conception. The proposed embryogenesis postulates failure of lateral mesoderm folds to migrate to the midline, causing the sternal and abdominal wall defects and failure of septum transversum to develop, causing defects in the anterior diaphragm and pericardium.<sup>3</sup>

### DEFINITION

The complete syndrome is characterized by two major defects: ectopia cordis and an abdominal wall defect

(most commonly omphalocele but gastroschisis can also be present). The other three defects of pentalogy of Cantrell are disruption of all the interposing structures: the distal sternum, anterior diaphragm and diaphragmatic pericardium. Incomplete expressions have also been reported. Toyama suggested the following classification of pentalogy of Cantrell.

- *Class 1:* Definite diagnosis with all five defects present.
- *Class 2:* Probable diagnosis with four defects present including intracardiac and ventral wall abnormalities.
- *Class 3:* Incomplete expression with various combinations of defect present including sternal abnormality.

Cardiac Echo was not done in our case. But in our case, hydrocephalus, spine, ear and hand deformities were present. So, our patient expressed incomplete form of pentalogy of Cantrell.

### PREVALENCE

Very less: 1 per 65,000 to 5.5 in a million live births.

### ULTRASOUND DIAGNOSIS

Diagnosis requires five criteria described by Cantrell, but incomplete variants have been described as in our case.<sup>1</sup> The most common intracardiac defects are atrial septal defect (ASD), ventricular septal defect (VSD) and tetralogy

of fallot (TOF). Intrauterine diagnosis of pentalogy of Cantrell is impossible before 12th week of pregnancy as herniation of bowel is a normal event in fetal development at this time, but after that ultrasound is useful method for diagnosis even in 1st trimester. If abdominal wall defect is present together with other abnormalities, specially ectopia cordis, one should consider pentalogy of Cantrell.

## PROGNOSIS

In a review of literature in 1972, Toyama reported a survival rate of 20%.<sup>2</sup> Three of five patients Cantrell reported in 1958 survived, but none of the five had true ectopia cordis. Prognosis may be related to the extent of ventral wall, sternal and cardiac defects.

## Associated Anomalies

Anomalies associated may be cranial or facial anomalies, club foot, malrotation of colon, hydrocephalus and anencephaly. Fetal echo is done to detect intracardiac abnormalities. Chromosomal analysis, as association with trisomy 18, 13, 21, has been reported.

## MANAGEMENT

In view of poor prognosis, termination of pregnancy can be considered if diagnosis is made before period of viability. After delivery, gastroschisis or omphalocele repair

should not be delayed. Repair of sternal, diaphragmatic and pericardial defects can be attempted at same time. Surgical correction is often difficult secondary to hypoplasia of thoracic cage and inability to enclose ectopic heart. Recognition and treatment of any intracardiac anomaly is important as congenital heart disease is a source of major morbidity in infants surviving the neonatal period.<sup>4</sup>

## CONCLUSION

Prenatal routine sonography in 2nd trimester may screen pentalogy of Cantrell. So, emphasis must be given to 2nd trimester ultrasound and offered to each and every patient.

## REFERENCES

1. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. *Surg Gynecol Obstet* 1958;107:602-614.
2. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. *Pediatr* 1972;50:778-792.
3. Haynor DR, Shuman WP, Brewer DK. Imaging of fetal ectopia cordis: roles of sonography and computed tomography. *J Ultrasound Med* 1984;3:25-27.
4. Fox JE, Gloster ES, Mirchandani R. Trisomy 18 with Cantrell pentalogy in a stillborn infant. *Am J Med Genet* 1988;31:391-394.